



TGM1 gene

transglutaminase 1

Normal Function

The *TGM1* gene provides instructions for making an enzyme called transglutaminase 1. This enzyme is found in cells that make up the outermost layer of the skin (the epidermis). Transglutaminase 1 is involved in the formation of the cornified cell envelope, which is a structure that surrounds skin cells and helps form a protective barrier between the body and its environment. Specifically, transglutaminase 1 forms strong bonds, called cross-links, between the structural proteins that make up the cornified cell envelope. This cross-linking provides strength and stability to the epidermis.

Health Conditions Related to Genetic Changes

lamellar ichthyosis

Many mutations in the *TGM1* gene have been found to cause lamellar ichthyosis, which is a condition that causes scaly skin that covers much of the body, and other skin abnormalities. Some *TGM1* gene mutations that cause this condition change single DNA building blocks (nucleotides) in the transglutaminase 1 enzyme. The most frequently occurring mutation (written as 877-2A>G) affects the way the gene's instructions are pieced together to form the enzyme and results in an abnormally shortened, nonfunctional enzyme. Other *TGM1* gene mutations result in a transglutaminase 1 enzyme that cannot function normally, is abnormally short, or is not produced. A lack of functional transglutaminase 1 prevents the formation of the cornified cell envelope, causing the skin abnormalities of lamellar ichthyosis.

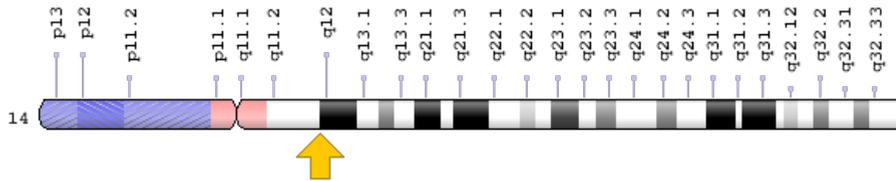
other disorders

In addition to lamellar ichthyosis (described above), *TGM1* gene mutations have been found to cause other forms of ichthyosis. In one type, called self-healing collodion baby, affected infants are born with a tight, clear sheath covering their skin called a collodion membrane. This membrane usually dries and peels off during the first few weeks of life, and affected infants often show near normal skin within a few months. Another type of ichthyosis, called bathing suit ichthyosis, is characterized by scaly skin that is limited to the trunk.

Chromosomal Location

Cytogenetic Location: 14q12, which is the long (q) arm of chromosome 14 at position 12

Molecular Location: base pairs 24,249,114 to 24,263,210 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- epidermal TGase
- ICR2
- protein-glutamine gamma-glutamyltransferase K
- TGASE
- TGase-1
- TGase K
- TGK
- TGM1_HUMAN
- transglutaminase-1
- transglutaminase 1 (K polypeptide epidermal type I, protein-glutamine-gamma-glutamyltransferase)
- transglutaminase K
- transglutaminase, keratinocyte

Additional Information & Resources

Educational Resources

- Molecular Biology of the Cell (fourth edition, 2002): Epidermal Cells Form a Multilayered Waterproof Barrier
<https://www.ncbi.nlm.nih.gov/books/NBK26865/#A4082>

GeneReviews

- Autosomal Recessive Congenital Ichthyosis
<https://www.ncbi.nlm.nih.gov/books/NBK1420>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28TGM1%5BTIAB%5D%29+OR+%28transglutaminase+1%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D>

OMIM

- TRANSGLUTAMINASE 1
<http://omim.org/entry/190195>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_TGM1.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=TGM1%5Bgene%5D>
- HGNC Gene Family: Transglutaminases
<http://www.genenames.org/cgi-bin/genefamilies/set/773>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=11777
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/7051>
- UniProt
<http://www.uniprot.org/uniprot/P22735>

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